Dowling-Degos disease

Dowling-Degos disease is a skin condition characterized by a lacy or net-like (reticulate) pattern of abnormally dark skin coloring (hyperpigmentation), particularly in the body's folds and creases. These skin changes typically first appear in the armpits and groin area and can later spread to other skin folds such as the crook of the elbow and back of the knee. Less commonly, pigmentation changes can also occur on the wrist, back of the hand, face, scalp, scrotum (in males), and vulva (in females). These areas of hyperpigmentation do not darken with exposure to sunlight and cause no health problems.

Individuals with Dowling-Degos disease may also have dark lesions on the face and back that resemble blackheads, red bumps around the mouth that resemble acne, or depressed or pitted scars on the face similar to acne scars but with no history of acne. Cysts within the hair follicle (pilar cysts) may develop, most commonly on the scalp. Rarely, affected individuals have patches of skin that are unusually light in color (hypopigmented).

The pigmentation changes characteristic of Dowling-Degos disease typically begin in late childhood or in adolescence, although in some individuals, features of the condition do not appear until adulthood. New areas of hyperpigmentation tend to develop over time, and the other skin lesions tend to increase in number as well. While the skin changes caused by Dowling-Degos disease can be bothersome, they typically cause no health problems.

A condition called Galli-Galli disease has signs and symptoms similar to those of Dowling-Degos disease. In addition to pigmentation changes, individuals with Galli-Galli disease also have a breakdown of cells in the outer layer of skin (acantholysis). Acantholysis can cause skin irritation and itchiness. These conditions used to be considered two separate disorders, but Galli-Galli disease and Dowling-Degos disease are now regarded as the same condition.

Frequency

Dowling-Degos disease appears to be a rare condition, although its prevalence is unknown.

Genetic Changes

Mutations in the *KRT5* gene cause Dowling-Degos disease. The *KRT5* gene provides instructions for making a protein called keratin 5. Keratins are a family of proteins that form the structural framework of certain cells, particularly cells that make up the skin, hair, and nails. Keratin 5 is produced in cells called keratinocytes found in

the outer layer of the skin (the epidermis). Keratin 5 is one component of molecules called keratin intermediate filaments. These filaments assemble into strong networks that help attach keratinocytes together and anchor the epidermis to underlying layers of skin. Researchers believe that keratin 5 may also play a role in transporting melanosomes, which are cellular structures that produce a pigment called melanin. The transport of these structures into keratinocytes is important for normal skin coloration (pigmentation).

KRT5 gene mutations that cause Dowling-Degos disease lead to a decrease in the amount of functional keratin 5 protein that is produced. A reduction in keratin 5 can impair the formation of keratin intermediate filaments. As a result, the normal organization of the epidermis is altered, leading to the development of different types of skin lesions. Additionally, a decrease in keratin 5 may disrupt the movement of pigment-carrying melanosomes into keratinocytes, where they are needed for normal skin pigmentation. This disruption of melanosome transport is thought to cause the pigmentation abnormalities seen in individuals with Dowling-Degos disease.

Some people with Dowling-Degos disease do not have an identified mutation in the *KRT5* gene. In these cases, the cause of the condition is unknown.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- dark dot disease
- DDD
- Dowling-Degos-Kitamura disease
- reticular pigment anomaly of flexures
- reticulate acropigmentation of Kitamura

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Reticulate acropigmentation of Kitamura https://www.ncbi.nlm.nih.gov/gtr/conditions/C0406811/

Other Diagnosis and Management Resources

- Cleveland Clinic: Skin Care Concerns http://my.clevelandclinic.org/health/articles/skin-care-concerns

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Skin--Abnormally Dark or Light
  https://medlineplus.gov/ency/article/003242.htm
- Health Topic: Skin Pigmentation Disorders
  https://medlineplus.gov/skinpigmentationdisorders.html

Genetic and Rare Diseases Information Center

- Dowling-Degos disease
  https://rarediseases.info.nih.gov/diseases/9775/dowling-degos-disease

Educational Resources

- Cleveland Clinic: Abnormal Pigmentation
  http://my.clevelandclinic.org/health/articles/abnormal-pigmentation
- Disease InfoSearch: Dowling-Degos Disease
  http://www.diseaseinfosearch.org/Dowling-Degos+Disease/2326
- Johns Hopkins Medicine: Skin Pigment Disorders
  http://www.hopkinsmedicine.org/healthlibrary/conditions/dermatology/skin_pigment_disorders_85,P00304/
- MalaCards: dowling-degos disease
  http://www.malacards.org/card/dowling_degos_disease

- Merck Manual for Health Care Professionals: Overview of Skin Pigment
  http://www.merckmanuals.com/home/skin-disorders/pigment-disorders/overview-of-skin-pigment
- Orphanet: Dowling-Degos disease
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=79145

Patient Support and Advocacy Resources

- American Skin Association
  http://www.americanskin.org/
- Children's Skin Disease Foundation
  https://www.csdf.org/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28dowling-degos+disease%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- DOWLING-DEGOS DISEASE 1
  http://omim.org/entry/179850

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